



FA FAMILY NEWSLETTER

#38

A Semi-annual Publication of the Fanconi Anemia Research Fund, Inc.

Fall 2005



Andy Eichenfield, the Camp doctor, and the Camp dog lead the walk to the pond for the Wish Boat launching.

Annual FA Family Meeting

A record 49 FA families, from as far away as Australia, Norway, and Germany, arrived at Camp Sunshine on Sebago Lake, Maine, in August for the 15th Annual FA Family Meeting. Those FA families who had attended past meetings were delighted to see one another again and instantly took up where they left off last year. The fears of new families were immediately calmed by the warmth of Camp Sunshine's welcoming volunteers, assisted by furry costumed bears and a chipmunk that

joined in the greeting, dispensing hugs and high-fives to the awestruck little ones.

The meeting was packed with wonderful recreational activities for the kids, from the toddlers to the teens. This was the first time that many FA patients had met another FA patient. That, alone, made Camp worthwhile. For siblings of FA patients, Camp presented a unique opportunity to share the experience of dealing with FA with other

continued on page 12

Two New FA Genes Isolated

Major advances in FA science were recently reported in the August 20, 2005 online edition of the prestigious journal, *Nature Genetics*. Leading laboratories have isolated two FA genes, *FANCF* and *FANCM*. The FA Research Fund has given generous support to these efforts.

FANCM, also called FAAP250, was isolated by A. Ruhikanta Meeti, et al., under the direction of Weidong Wang, PhD of the NIH National Institute of Aging. The protein associated with this gene is a member of the FANCF "core

continued on page 14

HIGHLIGHTS

New Therapies with Potential to Improve Transplantation Discussed	2
Chromosome Three Changes Seen as Warning Signal	2
Head and Neck Cancer in FA Patients	4
Harris Receives Lifetime Achievement Award.....	8
Family News	9
2005 Family Meeting at Camp Sunshine	12
Fundraising.....	19

New Therapies with Potential to Improve Transplantation Discussed

Margy MacMillan, University of Minnesota, brought hope and optimism to Camp Sunshine with her exciting presentation of four potential new therapies for FA patients which she and her colleagues, including John Wagner, have developed. These therapies are designed to reduce transplant-related toxicity, graft-versus-host disease (GVHD) and infections, and to improve immune recovery and survival.

1) Multipotent adult progenitor cells

One single multipotent adult progenitor cell derived from adult bone marrow can be cultured and expanded *in vitro*, and can retain the ability to differentiate into various tissues. In mouse models, these cells seek out areas of injury, suppress the host's immune system, and engraft, without causing tumor formation. It is hoped that these cells can be used in FA patients to regenerate and repair tissues following transplantation and could be a vehicle for gene therapy. The capacity of these cells to proliferate in different tissues throughout the body makes them particularly hopeful for a disease that affects all body tissues.

Researchers at the University of Minnesota are planning their first clinical trial of MAPC, to begin in January 2006. Only high-risk transplant patients (those with fungal or gram-negative infections or heavily transfused patients) will be eligible for this first trial which will test the safety of this approach.

2) Keratinocyte growth factor

In non-FA patients, keratinocyte growth factor reduces side effects of bone marrow transplantation, especially mouth sores. Researchers anticipate that this growth factor may enhance immune recovery and

decrease the risk of infections in FA patients after transplant. Discussions are underway with Amgen to make this product available to FA patients.

3) Mesenchymal stem cells

Mesenchymal stem cells are the honeycomb-like cells that support the bone marrow. They can be isolated from donor marrow and grown in the laboratory. When given at the time of transplant, they may speed engraftment, reduce toxicity and prevent graft-versus-host disease.

4) T Regulatory Cells

T regulatory cells are a subset of lymphocytes. They can be isolated



Margy MacMillan, MD

and grown from donor marrow, and can be infused at the time of transplant. These cells have been shown to reduce GVHD in mice. The first human trials are about to start at the University of Minnesota. Initially these trials will be in non-FA patients with the hope of bringing them to FA patients in the near future. ♦

Chromosome Three Changes Seen as Warning Signal

Families attending Camp Sunshine heard experts from two transplant centers speak of the significance of subtle changes in chromosome 3 in FA patients. If a gain is detected in the long arm of chromosome 3, Dr. Wolfram Ebell of Charité Hospital in Berlin believes this event signals a progression that will result in myelodysplasia or leukemia. Ebell believes proceeding quickly to transplant is necessary under these circumstances. Dr. Richard Harris of Cincinnati Children's Hospital agreed that concern about this clonal progression was consistent with his own experience.

Heidemarie Neitzel, PhD, of the Institute for Human Genetics, Charité, Berlin has published a peer-reviewed study of this phenomenon in the journal *Blood*. FARF recently funded a proposal to track patients in a number of centers internationally to determine whether systematic monitoring of this chromosomal change in the bone marrow and the peripheral blood is a reliable indicator for the timing of a stem cell transplant. ♦

IVF/Preimplantation Genetic Diagnosis

Renee Genovese, MS, Genetic Counselor, Reproductive Genetics Institute (RGI), Chicago, discussed preimplantation genetic diagnosis (PGD) as an option for FA parents at the August FA Family Meeting. PGD makes it possible to diagnose genetic disease in fertilized eggs/embryos *before* a pregnancy occurs. This procedure can also be used to test for HLA antigens, thereby determining if a pregnancy might provide a matched sibling donor for a later transplant.

In vitro fertilization (IVF) is done in conjunction with PGD. Parents can opt to come to Chicago for the entire procedure or work with a local IVF facility in addition to the Reproductive Genetics Institute in Chicago. RGI works with over 70 IVF centers throughout the country and outside of the United States.

Fertile couples go to a clinic

dealing with infertility problems.

The mother takes hormones, which stimulate her ovaries to produce multiple eggs. The eggs are retrieved, fertilized with the husband's sperm, and allowed to develop in a test tube. One cell from each embryo (called a blastomere) is removed on the third day following egg retrieval. This cell is tested for absence of the genetic disease and HLA antigens, if requested. With FA, there are 3 chances in 4 that a blastomere will be free from FA, and 3 chances in 16 that any one blastomere will be free from FA and will be an HLA-matched donor for an FA sibling. Removal of one blastomere cell from the embryo carries a less than 1% risk that the embryo will not develop.

Genovese stated that RGI takes great care to maximize the possibility that only healthy embryos are returned to the mother. In addition



Renee Genovese, MS

to testing one cell from the blastomere, cells called "polar bodies" from the egg are studied for "linked markers" that give assurance that the embryo(s) selected for transfer will be healthy. While no center will promise 100% accuracy, RGI quotes 98% accuracy.

continued on page 17

Transplant Results in Germany

Wolfram Ebell, MD, Charité Hospital, Berlin, reported on 195 FA patients who have been followed over the past two decades in Germany. While both transplanted and non-transplanted patients are living longer, those who have not under-



Wolfram Ebell, MD

gone transplant have generally lived longer than transplanted patients. Patient characteristics between these two groups, especially severity of disease causing a need to transplant, probably explain this difference.

Ebell emphasized the importance of identifying those patients who have a very poor prognosis without a bone marrow transplant. The presence of a subtle clonal abnormality on chromosome 3 is a powerful predictive factor. Germany has followed 18 patients with this abnormality, and the outcome has been grim. Once this clone appears, it does not go away but expands in the bone marrow, leading to myelodysplasia (MDS) and leukemia. These patients need to be transplanted well before the onset of leukemia.

Ebell has performed 17 unrelated

donor transplants at his center using fludarabine, low-dose busulfan, and a non-irradiation protocol. Of 17 patients, 11 survive. Survival was better in patients with aplastic anemia or myelodysplasia (10 of 14 survive) than in those with leukemia, where only one patient of three survived.

Ebell states that his own center waits longer than others to proceed to transplant. He believes that the window between the diagnosis of MDS and leukemia is large enough to locate an appropriate donor and proceed to transplant.

Of great concern is a 12-year-old patient who developed squamous cell carcinoma of the tongue very early after transplant. This patient did not have graft-versus-host disease, which is known to be associated with later malignancies. ♦

Head and Neck Cancer in FA Patients

David Kutler, MD, Department of Otolaryngology, Head and Neck Surgery, NYU Medical Center, spoke at Camp Sunshine on prevention, surveillance recommendations, and treatment of squamous cell carcinoma of the head and neck in FA patients. In non-FA patients in the United States, use of alcohol and/or tobacco is the main risk factor for these cancers, accounting for 85% of all diagnosed cases. Chewing betel nuts, certain viruses (human papilloma virus and Epstein-Barr virus), and genetic diseases such as FA also cause cancers of the head and neck. **Kutler strongly recommended that all FA patients refrain entirely from the use of alcohol or tobacco. In addition, FA patients should never be exposed to second-hand smoke.**

In FA patients, there is a 21%

incidence of these cancers by age 40. Age of onset is as young as 10. Kutler discussed 19 FA patients with squamous cell carcinoma of the head and neck. Most of these cancers appeared in the mouth (as opposed to more distant regions of the throat or neck); most were in either stage 1 or stage 4 at the time of diagnosis. Ten patients experienced a recurrence of the cancer, usually in the first year after removal of their tumor. Fourteen of nineteen patients died; eleven of these died because of their malignancy.

Kutler emphasized the importance of prevention of these cancers. In addition to refraining from exposure to alcohol or tobacco, **patients are advised to maintain good oral hygiene.** If a causal connection can be established between viral infections such as HPV and these cancers



David Kutler, MD

in FA patients, it would be advisable to immunize patients against these viruses. To date, immunization against HPV has not received final FDA approval.

continued on page 7

Endocrine Problems in FA Patients; Nutritional Needs Noted

Susan Rose, Professor of Endocrinology at Cincinnati Children's Hospital Medical Center, discussed the importance of identifying and treating endocrine problems in FA patients. Endocrine abnormalities



Susan Rose

are extremely common in FA patients; many problems are responsive to therapy that might improve growth and overall health.

Over the past three years, endocrinologists at the FA Comprehensive Care Center, Cincinnati Children's Hospital Medical Center, have conducted endocrine studies in 48 children with Fanconi anemia. Over a third of these children were small for gestational age. Sixty-seven percent (32 of 48) had short stature. Eighty percent of the FA children studied had abnormal thyroid tests, although many of these abnormalities were "mild."

Of the 32 children able to complete the oral glucose tolerance test (OGTT), 28 (88%) had abnormal insulin-glucose levels. Some made insulin but were not sensitive to their own insulin; others failed to make adequate insulin. Eighty-eight

percent had hyperglycemia or high blood sugar (47% had impaired glucose tolerance and 41% overt diabetes mellitus).

Rose emphasized the special nutritional needs of FA patients. All patients should avoid food with a "high glycemic index" or foods that rapidly raise blood sugar. These include concentrated sweets (such as pop tarts, colas, juice, sweet cereal); white bread, white bagels, white rice, potatoes, pasta; and jelly beans, life-savers, corn chips, pretzels, and rice cakes. Foods with a "low glycemic index" are greatly preferable. These include nuts, peanut butter, beans, whole grain bread, brown rice and whole-wheat pasta; fruits and vegetables; cheese, milk, meats or poultry; peanut M&Ms, Snickers (nuts or protein causes foods to be digested more slowly, preventing the rapid conversion to sugar). ♦

FA Treatment Update from Cincinnati

Participants at Camp Sunshine again welcomed Franklin Smith, MD, of the Cincinnati Children's Hospital Fanconi Anemia Comprehensive Care Clinic. Smith described ongoing research projects related to FA. Among notable developments are the following:

- In the normal population, 50% of people have a gene called *GSTM* and in 50%, this gene is missing. FA-C children who lack this gene all have early onset bone marrow failure.
- FA patient marrow stem cells die in part because they develop high levels of a protein called tumor necrosis factor (TNF), which is toxic to stem cells. However, a currently licensed drug, etanercept, is known to neutralize TNF. Cincinnati will shortly open a study for children just starting to develop low counts to see whether this drug, which is taken as an injection once a week, can preserve the participants' stem cells.
- Androgens, which have troublesome side effects for some patients, also can delay marrow failure for about two years. About 50% of FA patients respond to androgens. Cincinnati has opened a new study testing an androgen called oxandrolone. This drug may have fewer side effects and work better. Six patients have enrolled in the study; four have responded positively to this drug to date.
- Matched sibling FA transplants at Cincinnati have resulted in a high success rate. Of the last 35 sibling donor transplants performed at this center, 88% of the children are alive and well with normal blood counts.
- In order to reduce the risk of cancer, FA children receiving sibling donor transplants in the future will not receive radiation. They will receive a special antibody (Campath) to help new cells grow and to prevent graft-versus-host disease.
- Cincinnati has begun doing pre-transplant chemotherapy on FA children who develop leukemia or severe MDS with abnormal chromosomes (e.g., monosomy 7). Of the six children treated with this FLAG (fludarabine/cytarabine and GCSF) therapy, three were cleared of the disease and are alive. ♦



Franklin Smith, MD

Patrick Kelly Gives Gene Therapy Update



Patrick Kelly, MD

Dr. Patrick Kelly gave Camp Sunshine attendees a report on the current gene transfer trial at Cincinnati Children's Hospital. This trial is currently open to FA-A patients only. Of the first three patients treated, one had had marrow frozen five years ago. The process of freezing, thawing, and culturing these cells destroyed too many cells for re-infusion. Two patients were infused with fresh gene-corrected stem cells. One briefly showed signs of corrected cells in the blood; the other did not. Investigators hope that examination of the bone marrow in coming months will indicate the presence of corrected cells.

A major challenge to gene therapy is the fragility of FA stem cells. Many

cells are lost when they are manipulated outside of the body. New methods for freezing and thawing these cells are currently in use, which should improve their viability. Because of these advances, Kelly emphasized the importance of early stem cell harvests from patients in order to obtain adequate numbers of cells for successful gene therapy.

A gene therapy trial for FA-G patients is pending and scientists are preparing for a later trial of FA-C patients. Patients who wish more information concerning these trials should contact Robin Mueller, RN at (513) 636-3218 or <RobinMueller@CCHMC.org>. ♦

Ear and Hearing Problems Common in FA

Hearing disorders in Fanconi anemia patients are far more frequent than earlier medical literature has reported. Dr. H. Jeffrey Kim of the National Institute on Deafness & Other Communication Disorders of the NIH reported on results of a recent comprehensive examination of 12 FA patients for ear abnormalities and hearing loss.

Almost 50% of the patients studied had mild and moderate hearing loss. Most of the hearing loss is “conductive” in nature while the inner ear and hearing nerve function are mostly unaffected. Most of the patients (even those with normal hearing) had structural abnormalities in the outer and middle ears. These common structural changes include small eardrums and ear canals, and malformed bones in the middle ear.

Kim advised that FA patients undergo a thorough hearing evaluation by expert audiologists soon after the diagnosis of FA. If necessary, computerized tomography (CT) imaging can help diagnose potential problems. Mild to moderate hearing loss makes it difficult to detect sounds with background noises; decreases one’s responsiveness to work and school environments; and

makes it difficult to hear certain sounds. Therefore, early auditory rehabilitation and speech therapy may be critically important to prevent the possibility that undetected hearing loss will interfere with normal speech and language development and overall school performance. Management options for hearing loss fall into three major categories, depending on the cause and extent of hearing loss and underlying health status of FA patients:

1. Auditory amplification through hearing aids;
2. Surgical correction to widen the ear canal and to address middle ear bone problems; and
3. Implantation of a hearing device called “bone anchoring hearing aids” (BAHA®). ♦



Jeffrey Kim, MD

Gynecological Issues Reviewed

Alicia Armstrong, MD, of the Reproductive Biology and Medicine Branch, NICHD, NIH, reviewed gynecological and reproductive issues facing FA patients. For women, these include later onset of menses and premature menopause. Many experience irregular periods and excessive menstrual bleeding. While women with FA have a shortened reproductive life and decreased fertility, they can become pregnant. Pregnancy carries increased risks of C-section, lower blood counts often requiring transfusions, low birth weight, and premature delivery, but these risks are medically manageable.

Transplantation drastically reduces a woman’s ability to conceive a child. Armstrong discussed embryo or egg freezing prior to transplant and ovarian shielding strategies as possible ways to overcome this complication. (Embryo freezing is more effective than freezing eggs). One strategy to preserve ovarian function is administration of the drug Lupron prior to transplant. This drug inactivates the ovaries, making it more likely that ovarian function can be maintained following transplantation.

Finally, Armstrong noted the importance of screening the FA female patient for the increased risk of vulvar and cervical cancer. Regular gynecological exams including Pap smears and colposcopies (examining the vagina and cervix by means of a magnifying lens) should begin from onset of menarche, sexual activity or at age 16. Breast cancer screening should include self and physician examinations.

Men with FA also have decreased fertility; many have zero sperm count. If sperm counts are not zero, *in vitro* fertilization or freezing sperm may increase the likelihood of fertilization. ♦



Alicia Armstrong, MD

Practical Communication Tips for Hearing-impaired Children

- Help your child develop the habit of watching the speaker.
- Encourage your child to ask the speaker to repeat information that was missed.
- Reduce or move away from background noises.
- Do not emphasize pronunciation as you speak. Speak clearly and slowly.

Head and Neck Cancer in FA Patients

continued from page 4

Regular surveillance is crucial to identifying head and neck cancers early. Surveillance should begin at age 10, and must be conducted by a physician with experience in identifying and treating these cancers. Evaluation should include examination of the mouth, and flexible fiberoptic scope of the throat every six months. Conditions such as leukoplakia (a white plaque which can't be scraped off) and especially erythroplakia (a red plaque) are precancerous conditions and should be followed closely. **Erythroplakia and persistent leukoplakia must be biopsied.**

Treatment of these cancers in FA patients is complicated by patient sensitivity to radiation and chemotherapy. Kutler discussed eight patients who underwent radiation therapy. All six who received this therapy after surgery experienced severe complications; three could not complete the treatment and two died of radiation exposure. Early detection and surgery remain the best approaches to these cancers; radiation and chemotherapy should be used with great caution in very limited circumstances. Post-treatment surveillance is crucial. ♦

Your FA Research Dollars at Work in 2005

Through the end of September, 2005, the Fanconi Anemia Research Fund awarded \$853,184 in research grants to the following projects:

- Investigator: Ruud Brakenhoff, PhD, Vrije Universiteit Medical Center, Amsterdam, The Netherlands
Title: *Genetic Progression of FA Squamous Cell Carcinoma and Development of a Non-invasive Screening Method for Precursor Lesions*
Amount: \$265,800
- Investigator: Laura Haneline, MD, University of Indiana School of Medicine
Title: *Preclinical Analysis of Potential Therapeutic Agents Targeted to Enhance FANCC-/- HSC Function*
Amount: \$176,813
- Investigator: Susan Rose, MD, Cincinnati Children's Hospital Medical Center
Title: *Thyroid Hormone in Children with FA*
Amount: \$47,687
- Investigator: Patrick Kelly, MD, Cincinnati Children's Hospital Medical Center
Title: *Gene Transfer for Patients with FA Genotype A*
Amount: \$101,196
- Investigator: Johan de Winter, PhD, Vrije Universiteit Medical Center, Amsterdam, The Netherlands
Title: *A Knock-Out Mouse for Fancm*
Amount: \$96,000
- Investigator: Holger Tonnies, PhD, and Heidemarie Neitzel, PhD, Institute for Human Genetics, Humboldt University, Berlin, Germany
Title: *Multi-Center Study for Correlating the Clinical Data with Clonal Aberrations in Mononuclear Peripheral Blood Cells of FA Patients*
Amount: \$101,688
- Investigator: Duane Lehtinen, PhD, and Thomas Hollis, PhD, Wake Forest School of Medicine
Title: *Crystallographic Studies of the Fanconi Anemia L Protein*
Amount: \$64,000

Harris Receives *Lifetime Achievement Award*

The Board of Directors of the FA Research Fund voted to bestow the Fund's *Lifetime Achievement Award* on Richard Harris, MD, for his many years of dedication to FA patients.

The award was presented in August by Dave Frohnmayer, vice-president of the Board of Directors, at the FA Annual Family Meeting at Camp Sunshine. The site was particularly fitting because of Dr. Harris's faithful attendance at this meeting each year, during which he has presented his bone marrow transplant results and visited with FA families in the informal atmosphere of the Camp.

The Board of Directors has acknowledged the exceptional contribution of Dr. Harris in the area of bone marrow transplantation for FA patients. Because of his work, the odds of FA patients surviving transplants have increased and their quality of life post-transplant has improved. Equally important, the Board acknowledged that Dr. Harris has excelled in establishing strong relationships with his FA patients and their families. He clearly cares about his patients, and the Fund is deeply grateful for his commitment to them and to our cause. ♦



Richard Harris, MD receives Lifetime Achievement Award from Dave Frohnmayer.

Regional Meeting Held in New York City

The Fanconi Anemia Research Fund convened a Regional Meeting at the Millennium Hotel in New York City on Sunday, June 3, 2005. Twenty-seven FA patients and family members attended. The event featured presentations by a number of experts in FA medical care. Farid Boulad, MD, Memorial Sloan-Kettering Cancer Center (MSKCC), explained hematopoietic stem cell transplantation for FA patients, and Bhuvanesh Singh, MD, also of MSKCC, presented on the squamous cell carcinoma of the head and neck that affects so many FA patients. Speakers in the afternoon included Al Gillio, MD, who spoke on the medical issues faced by adult FA patients, and Daniel Smith, MD, who presented on gynecological issues. Both Gillio and Smith are from Hackensack University Medical Center. Arleen Auerbach, PhD, The Rockefeller University, discussed complementation group testing.



FA patients and family members listen to Farid Boulad discuss transplants.

FA Research Fund Fundraising Team Leaders Audrey Barrow, Randy Bloxom, and Peggy Padden made an excellent presentation to encourage FA families to raise money for much-needed FA research.

The day concluded with a "Coping with FA" session led by Nancy Cincotta, MSW, Mt. Sinai Medical Center. As always, this opportunity to get to know other FA families better was a highlight of the meeting. ♦



Living with Fanconi Anemia

by Paula Ceresa-Guidara

My name is Paula Ceresa-Guidara. I am married, and we have one child who is 12 years old. I am 51 years old and have Fanconi anemia. I have been teaching elementary school for the past 29 years.

I do not know my complementation group or my mutation. This has not been of importance to me because I have not needed this information to help with my care. I have never had any marrow or blood problems, but I have had numerous surgeries in my mouth for cancer and

mild to moderate to severe dysplasia. Additionally, I will be undergoing minor surgery in the near future for some gynecological problems.

I try to maintain good health by washing my hands frequently and getting an ample amount of sleep although, being a night owl, I hate to go to bed! I eat a healthy diet that includes all the food groups. I take supplemental calcium and vitamins. I don't smoke and only drink alcohol on occasion. I have a wonderful

continued on page 17

Our Experience in a Clinical Trial

by Kristina King

We first heard of the gene therapy clinical trial at Cincinnati Children's Hospital Medical Center at our complete FA evaluation in January, 2004. During that visit and when we arrived home, things looked grim for FA kids. We talked about all the options. Along with Jo and Jacy, David and I decided that we really wanted to give the trial a try. When we asked the girls, they both responded, simultaneously, "If it does not help us, maybe we can help the other kids." That is when we decided to move to Cincinnati. There would be so many appointments and close monitoring that we could have never participated in the trial from Kansas.

In June, 2004 we moved and began a series of appointments to discuss the procedures, risks, new protocols, and FDA status. Things kept moving forward and getting better. Again, as we were ready to sign the

final papers, we asked the girls if they still wanted to participate in the trial, and they responded the same. They had to take tests to assure they knew all they needed to make the decision.

The advantages of this study are that we are improving the chances of a cure, testing whether it is a safe study, and allowing the doctors trying to find a cure the learning tools to proceed with greater knowledge. Disadvantages were none. Jo did want to say one thing for this article: "Remember not to get your hopes up, because it is not yet a cure."

We would and are planning to participate in the trial again, as many times as they will allow us. We feel that it can't hurt to try and, the more



Jo and Jacy King

the doctors learn from each case, the closer to saving lives we will be. The girls always say, "We may not save our lives, but we will help save others and, besides, it is cool that we are making history." ♦

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Ty Perkins

by Karen Magrath

August 22, 2005, Day 61, and barely a third of the way through our 6-month stay in New York City. That's where we are in the transplant process for our son, Ty.

The journey for me has been emotional: both draining and exhilarating. From the nagging worry that seems to accompany parenting a child with FA, even when things are good, to the utter realization in a facility such as this that things could be so much worse. From the wonderful friendships made and support gleaned within the FA community to the heart-wrenching anguish and discouragement I feel with each loss and every set-back. From the relative health that steroids and anadrol afforded Ty for 6 years to his rapidly deteriorating condition just prior to transplant. From the disappointment of finding out only weeks before transplant that our 10/10 donor was unavailable to our relief at finding out that we would proceed with a healthy 8/10 donor just weeks later than originally scheduled. From the moment the donor's cells arrived in Ty's room to 10 minutes later when the entire transplant was completed. From the horrible side effects of chemo and radiation to the day Ty's counts started coming in and his ravaged body started healing. From the day Ty was finally discharged from the hospital to the realization that our journey was only just beginning. From being separated by so many miles and so much time from my daughters and husband to racing home from Clinic each day to see them on the webcam, hear about their day, and listen to bed-time prayers. From the day we found out Ty's CMV (cytomegalovirus) was rearing its ugly head to TODAY when we found out his CMV is



Ty Perkins

again negative. From everything Ty has taught us through his bravery and laughter to what we've been able to learn only through faith. From the experience we realized life would never quite be the same nor mean the same thing for us. Draining and exhilarating all at once.

Ahhhhh, blessed we are (said in the infamous "Yoda-ese" Ty will be so pleased I snuck in here). We're taking one day at a time. ♦

Editors' Note and Disclaimer

Statements and opinions expressed in this newsletter are those of the authors and not necessarily those of the editors or the Fanconi Anemia Research Fund. Information provided in this newsletter about medications, treatments or products should not be construed as medical instruction or scientific endorsement. *Always consult your physician before taking any action based on this information.*

Find Answers

by Diane and Mark Pearl

As parents we will do anything for our children. As FA parents we are destined and determined to Find Answers. Quality of life is what you live for, most of the time just to get through each day. The following tips may help other parents alleviate small frustrations.

On a trip to Camp Sunshine in Maine, we discovered the Golden Access Passport (www.nps.gov/fees_passes.htm) that provides free entry into all National Parks in the US. This pass is valid for life and is available to individuals with special needs. Our FA children qualify. The pass allows your party in the "shorter" pass line, which can save hours at a popular park such as the Grand Canyon during peak times. We have used the kids' passes around 10 times, and now they think we should visit every national park in the country.

Similarly, you can request a "special needs pass" at theme parks such as Disney and Six Flags. Visit or call ahead to Guest Relations to request this pass. Prior to making the trip, contact your doctor and ask him or her to prepare a letter stating your child has FA, should not spend prolonged time in the sun, has a compromised immune system, or whatever condition applies to your child. This pass, usually a wristband, will allow you to use special entries and exits, thereby avoiding crowds and dangerous germs. Many times the pass will also provide for special seating at the various shows.

Getting to theme parks can present challenges. Several organizations including Dream Factory (www.dreamfactoryinc.com) and Make a Wish (www.wish.org) can provide airfare and hotel accommodations to defray the costs

continued on page 11

Attending Our First Family Meeting

by Janice Pless

Our first experience at Camp Sunshine this year can be summed up with one word: AWESOME! That's what our daughter, Julia, has been telling everyone who asks her about Camp. Our older daughter, Victoria, feels the same way and definitely wants to return next year, even though it means 12 hours of driving.

We all felt anxious about the first day. We didn't know what to expect.

Find Answers

continued from page 10

of a special trip for a critically ill child. Re-circulated air on commercial flights can also be a concern for children with poor immune systems. Angel Flight (www.angel-flight.org) is a group of pilots who donate their time and airplanes to take children and families on special trips and/or medical visits in the United States.

In addition to these special programs, we have had positive experiences with airline managers who have been willing to reduce otherwise prohibitive fares, waive fees, allow frequent flyer usage on blackout dates and make seats appear out of nowhere. Security post-9/11 has been increased at most airports. A letter from a doctor explaining that your child has FA can help your family avoid the crowded, germ-overloaded lines and get through much faster. Ask to board early to avoid the crowd during general boarding. This provides the opportunity to wipe down the germ-filled seats, trays and arm rests.

Remember that FA stands for Find Answers: it never hurts to ask. The worst thing that can happen is that you can be told "no." ♦

I personally was feeling very ambivalent. Did we really need to come here at this time? We had acquired most of the medical information about FA from my husband attending the clinical meetings, reading the publications put out by the FARF, and talking to FA medical specialists two years ago when we were faced with the diagnosis. At the time, Julia was in complete bone marrow failure, and we had to proceed to transplant as soon as possible. Now that she was post-transplant and stable, perhaps coming to Camp wasn't a priority for us. What could we gain?

Camp Sunshine is a very peaceful, beautiful, and serene place. So, the first thing we gained was a sense of relaxation. We all immediately felt comfortable and secure. I didn't think twice about who was caring for my girls because all the volunteers and staff were so warm, friendly, and kind. I just trusted them to take good care of my kids. Their days were filled with new friends, adventures, and loads of fun activities. They loved every single minute.

The first day for the adults was very fast-paced and overwhelming, especially emotionally, and this continued for three days. I am very grateful for the opportunity to listen to all the presenters. There is always something we can learn to help our FA children medically. We also appreciated hearing about the progress being made in FA research. This gives us hope for a positive future.



Julia Pless and Tom, the camp counselor

Finally, I realized that the most important gain of all was the support and understanding of people who knew exactly how I felt and what I was going through: a new family, my FA family. I had a safety net I could fall into and be caught!

Everyone comes to Camp with a different story about their experience so far with FA. Everyone is in a different place in dealing with FA. But, we all share in the realities and tragedies of living with FA every day. My husband said he felt "refreshed" after having attended Camp Sunshine, and I feel the same way. Camp has given us a brief respite from which we have come away feeling empowered and inspired to continue with life, knowing that we have this special family to come "home" to next year. ♦

Enjoying the 2005 Family Meeting at Camp Sunshine

Annual FA Family Meeting

continued from page 1

siblings. The kids all reveled in fun, from the Masquerade Ball to the Talent Show, the climbing wall, the putting green, the camp-out, and the campfires.

For the parents and adult FA patients, the Family Meeting provided an exceptional opportunity to hear from experts about treatments for and research into the many medical challenges that face FA patients. Hematologists discussed bone marrow transplantation, and oncologists discussed the high risk for head and neck and gynecological cancers in FA. Other professionals presented information on gene therapy research, ear and hearing problems, pre-implantation genetic diagnosis, and endocrine problems in FA. Although the information was exceedingly welcome, it was also overwhelming and often frightening. Fortunately, Nancy Cincotta, MSW, made herself available through many “coping with FA” sessions, so that parents could debrief what they heard, express their concerns, and receive support from Nancy and one another through this journey that is FA.

Attendees evaluated the meeting and made many constructive suggestions. All expressed their appreciation at being able to attend the meeting. ♦



Camp Sunshine Chipmunk and Bear welcome Amelia Bennett to Camp Sunshine.



Father and sons prepare to launch their Wish Boat.



Michelle Ploetz and “Pooh”

"Hard to see how this could be improved! It seems just ideal and exceptionally well run and welcoming."



Fast friends.

"I love Camp Sunshine! It is a great place to have this family meeting. The kids had a great experience. We look forward to coming back."

"This is an excellent location. . . . The entire arrangement in its nutshell time period was excellent."

"I appreciate the event. Thanks!"



FA parents and adults pay rapt attention to a medical presentation.



Donna Boggs, Maria Godwin, and Kayla Lackey at the Banquet

"Love to be here, educational and exciting. Good to see other parents and kids struggling with the same things as yourself. It made us happy, too, in the way that there are so many people helping and supporting patients with FA. The volunteers are exceptional and so is the Camp Sunshine staff!"



FA teens and friends enjoying each other's company.

Grandma and Grandpa Go to Camp

by Jeanne and Stuart Altman

Going to Camp Sunshine for the Family Meeting felt in advance like a step into the great unknown, with perceived benefits but also great risks. Our 2-year-old granddaughter Nina's diagnosis was only a few months old. Jeanne's initial thought was that Nina's parents, Rachel and Tyler, probably wouldn't want to consider Camp until another year. Dr. Blanche Alter strongly urged otherwise, and we offered to help Rachel if she decided to go to Camp with the kids and Tyler wasn't able to. Very shortly, they were all signed up for Camp. We both planned to go, particularly to be available for help with Nina, who had never had a day care experience, and her 8-year-old sibling Benjamin, who might react with great difficulty to this potentially overwhelming experience.

The kids were the first great surprise: instant success! Nina was in the wonderfully nurturing environment of the infant group, and Benjamin

thrived with the many wonderful age-based camp activities with both FA kids and "unaffected" siblings. By the end of the first day, he had planned what he'd be able to do next year when he'd be in the group that could learn archery, use the climbing wall, and have one overnight campout! Because Nina and Benjamin were so well cared for, we were able to attend most of the lecture presentations.

We had previously devoured most of the great FARF newsletters and other publications and had also read a lot of the technical literature. But the presentations were still valuable, as reviews, updates, or fresh information. And, the special value of these presentations was the ongoing accessibility of speakers for individual or



Nina Morrison and her parents having fun at Camp

small group discussions. What a unique resource, especially for parents! Other highlights included the high level of energy that both researchers and families seemed to draw from each other. We enjoyed the relaxation of an atmosphere in which one didn't need to explain one's situation to anyone. The same was true for the kids, whether sibling or FA: the natural and planned opportunities for kids to open up and, also, to develop healing perspectives for their family situation. It's difficult to say enough good about the caring and energy that were brought by the Camp Sunshine volunteer and permanent staff. We might summarize our impressions of the FARF and Camp staff in three words: thoughtful, compassionate, and informative.

If we sound like cheerleaders, it is perhaps because, as tough, exhausting, and sobering as the experience was, it was not the trade-off we imagined—it was powerfully good! Knowing the many reasons, fears, and exhaustion that might lead others to hesitate about attending Camp, we can't encourage them too strongly to do so. ♦

Two New FA Genes Isolated

continued from page 1

complex" that includes the A, B, C, E, F, G and L proteins, and which is essential for the monoubiquitination of the FANCD2 protein. This mechanism, still poorly understood, is crucial for recognition and response to DNA damage. Other organisms have similar functions to *FANCM*, making this gene of significant scientific interest.

The FANCI protein (also called BRIP1 and BACH1) was isolated as a previously discovered DNA-unwinding enzyme (helicase) in studies by Orna Levran, et al., in the New

York laboratory of Arleen Auerbach, PhD and Marieke Levitus, et al., in the Amsterdam laboratory of Hans Joenje, PhD. This J protein acts independently and downstream of the FANCD2 "core complex" and is implicated in the repair of damaged DNA strands.

While at least one more FA gene, *FANCI*, remains to be isolated, these recent developments advance FA science; make it clear that this science is closely associated with understanding cancer generally; and hasten the time when the crucially important processes of DNA repair biology are understood and harnessed for therapeutic purposes. ♦

Attending the Family Meeting

by Rachel Grossman

Despite the fact that we have been living with this disease for so long, this was our first time at Camp. Jacob was transplanted three years ago this past July. He continues to have many significant issues that are unrelated to transplant. He eats solely with a g-tube, will eventually need a new kidney, and has a myriad of other issues. We didn't go to Camp in the past for many reasons, which included some resistance from my husband. We came as a family this year and found that we are not alone. There are others just like us, post-transplant with continued issues.

We arrived at Camp to meet a whole host of wonderful people who all want the absolute best for their children. There was fun the first night with a get-to-know-you activity. It was a good taste of fun before the emotions started the next day.

On Saturday, we sat through a session for post-transplanted families. Several families who had attended Camp before noted that it was nice to see so many post-transplant families and their children. We were able to express our current feelings. Listening to others was invaluable during that session.

Talia, our eight-year-old daughter, sat with other siblings and discussed how she felt living with a brother with FA. Jacob was mad at first that he had to be with other FA children, but then enjoyed the weekend so much that he discussed his experience at school. He doesn't discuss his feelings much, but he ended up feeling at home at Camp.

Because we were post-transplant, we did not attend all of the sessions. We sat through both of the cancer sessions, and I felt that the floor was



FA parent Alan Grossman in a great karaoke performance at the Family Meeting!



Talia and Jacob Grossman

going to drop out from underneath me. My head was swirling, even though I only received part of the information that new families learned. I felt overwhelmed with the information; yet, I was able to see comfort in the families around us.

It was difficult to see children affected with this disease cry and worry about their future and that of their siblings. It was overwhelming to hear of the plight of others, yet comforting at the same time because we are all in the same "club" together. Despite the emotional roller-coaster, it was worth every second of the time that we spent at Camp and, the minute that we left Camp on Monday, we were already discussing next year. We have found lasting relationships that will be valued for years to come. We look forward to our future Camp experiences. ♦

In Memory of Emily Elaine “Firecracker” Clark

by Jeanette Clark

I am honored to write about a truly amazing person, my cherished daughter, Emily Elaine Clark. How can you put into a few paragraphs the “firecracker” personality of such a spirited person? From the day she was born, February 5, 1994, to the day she graduated, May 1, 2005, we all—family, friends, strangers alike—have been blessed with the on-the-go, spunky, let-nothing-stop-you personality in Emily. Her friends ranged in age from toddlers to the elderly. No matter. She accepted people for who they were. We were blessed in knowing that, although she was tiny and had to monitor foods, nothing could stop her. Size didn’t matter: the bigger the kids, the more she wanted to deal with them.

We didn’t hear of FA until April 20, 2004, when Emily wanted to try growth hormones; her friends were really getting ahead of her. On May 20th, FA was confirmed. Looking back, I realize we were fortunate to have Emily with us far longer than the statistics say. I noticed bruising when she was five years old, and her California tan stopped when she was eight. No one knew why.

Fanconi anemia was a tremendous shock and a challenge. The challenge was to learn about FA and to continue living a normal life without letting others focus on FA before they focus on Emily. We defied the statistics and stared FA in the face. I truly believe that Emily knew her physical life was ending, and she did everything she could to help us cope. She knew things that I can’t explain. One thing she told me was that I named her Emily, instead of Daniela Rose (dear friends are Danielle and Rose) because she’s “Em” for *FANCM*. I am now stunned to learn



Emily Elaine “Firecracker” Clark

about the recent scientific findings of exactly that.

From her natural operatic rendition of “Over the Rainbow” to her artistic drawings, flute playing, drama, and love of shoes, we will always remember her as “Our Little Angel.”

In her last weeks, she wanted Kelly Clarkson’s CD “Breakaway.” I heard it and cried. She was so aware and made the most of life. The day after she passed away, we heard all of her favorite songs on the car radio and saw the most beautiful full rainbow ever.

Thank you, Emily, for sharing your life with us. We’ll always be together and, until we see you again, we’ll honor your memory and keep you near. You’ll never be forgotten. We wish all the FA families courage, love and encouragement. ♦

In Loving Memory

Davaan McDuffie
12/16/02 – 4/6/05

Emily Elaine Clark
2/5/94 – 5/1/05

Leardon Keleher
2/11/62 – 5/30/05

The Vangel Family

by Mike Vangel

My wife, Beth, and I have two children, Amy (14) and Dennis (8). Both Amy and Dennis have Fanconi anemia. We had never heard of the disease until Amy was diagnosed with it at the age of four, when she had very low platelet counts. For almost 7 years Beth and I gave Amy two injections every other day of *Epogen* and *Neupogen* to keep her blood counts up, until finally she was not getting any benefit from the drugs. She needed to go to transplant.

Even with the millions of people in the bone marrow registry, we could not find a marrow match. Three years ago, on September 6, 2002, Amy had an unrelated, 5/6 HLA matched bone marrow transplant at Fairview University Hospital in Minneapolis. It was a hard decision to make, but Amy is a spunky kid and she is doing well now. Her recovery, though, took well over a year.

It is hard to look at Dennis, knowing that a transplant is almost certainly in his future. We have seen firsthand the rigors of Amy's transplant and the subsequent complications that can arise from a lengthy



Amy and Dennis Vangel

hospital stay. Even with the improved outcomes, we've seen other families who have lost children while going through the transplant process. We beat the odds once: can we do it again? We hope that when it comes time for Dennis to be transplanted, it will be a less painful and less lethal treatment. And, we hope that, for both of our children, medical research can find a way to treat and possibly prevent the solid tumor cancers of the head and neck that FA patients are 700% more likely to develop than the general population. ♦

Living with Fanconi Anemia

continued from page 9

family that is extremely supportive in all aspects of my life. I try to live my life by not dwelling on the negative and taking care of any health problems at hand and then moving on to enjoy all the good that life has to offer.

Sometimes though, even when you feel like you are doing all the right things, something goes wrong and you start having health problems. What has worked for me is to keep close watch for any changes in

or on my body and immediately report them to the doctor. I get check-ups about every three months with my gynecologist, oncologist, dentist, and general practitioner so that when I, my doctor or dentist sees anything suspicious, it can immediately be evaluated or the area in question can be biopsied, if necessary. I often have my doctors or dentist confer with a Fanconi anemia expert. The sooner you can discover a problem, the better chance you have to make a good informed decision about your health. That will help you to have a better quality of life. ♦

IVF/Preimplantation Genetic Diagnosis

continued from page 3

RGI has performed PGD for 110 different genetic conditions. A total of 447 patients have gone through 756 fertility cycles, and embryo transfer occurred 663 times. RGI has transferred 1290 embryos, resulting in 244 pregnancies. Thirty-five percent of embryos transferred resulted in pregnancy. To date, 212 babies have been born.

Seven FA families have worked with RGI to achieve a healthy pregnancy. These families experienced 18 IVF cycles. In total, twelve embryos were transferred to the mothers resulting in five pregnancies. Four healthy babies have been born (including one set of twins); one mother is still pregnant, and one pregnancy resulted in a miscarriage. The average success rate for FA parents has been ~25% per cycle. Much depends on the age and fertility characteristics of each couple.

Preimplantation genetic diagnosis is expensive, and insurance is unlikely to assume the cost. After an initial set-up fee (\$3,500-\$5,000 if only the health of the embryo is considered; an additional \$4,000 if the study will include HLA testing), the cost is approximately \$20,000 for each reproductive cycle.

Renee Genovese can be reached at:

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2825 N. Halsted
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Fax: 773-871-5221
Email: rgi@flash.net
www.reproductivegenetics.com ♦

One of Two in a Million

by Ben Murnane

I usually don't read this newsletter. The collection of tearful eulogies and movingly hopeful essays it contains is often too much heartbreak to take, for someone still living with the daily burden of having Fanconi anaemia.

I was diagnosed with FA in 1994 at the age of nine, too young to contemplate the tough times that lay ahead. I was prescribed the steroid oxymetholone and had a Broviac catheter inserted into my chest. But, all I knew was that I got to stay at home and watch cartoons while the other kids went to school, since my blood counts were dangerously low and I had to be kept in semi-isolation.

My troubles with FA came and receded over the next six years, like the tide lapping against the shoreline. But, hardly a day went by when I didn't feel shot, fatigued to the point of utter exhaustion. In the summer of 2000, I learned that the prolonged use of oxymetholone was causing growths to form on my liver: I was to be weaned off the drug immediately. I became dependent on blood transfusions then and spent some months injecting myself with that curse of the cycling profession—EPO—before I had to decide whether to undergo a bone marrow transplant. Without a BMT, I would certainly die within two years. But if the transplant proved unsuccessful, I could be dead within a few months. Moreover, I had to choose whether to have my sperm frozen, as after the radiotherapy necessary for the transplant, the chances of my being able to father a child naturally (already dramatically reduced by FA itself) would surely be devastated completely. These didn't seem like the kind of decisions I should have to make at fifteen years of age.

Between the summer and autumn of 2001, I spent twelve weeks in the transplant unit of St. John's Ward, Our Lady's Hospital for Sick Children, Crumlin, Dublin. The day of my release from hospital is one that still burns in my memory. It is a day, I'm sure, that everyone reading this recalls—though for a radically different reason than mine: September 11, 2001.

Since that date, the path of my recovery has not been entirely unblemished; yet, it has been smoother than anything that went before. After missing a year of secondary school, I went back and did my final exams. I'm currently attending college, studying drama. For now, at least, I'm free of the extreme effects of Fanconi anaemia. And, today, death at the hands of this illness seems a wholly conquerable prospect.

The above account could barely even be described as an outline of my experiences with FA—a disease which does not define me, but is as much a component of who I am as any aspect of my life. There's so much more I want to tell, so much more I want to share. That's why, for the past year and a half, on and off, I've been trying to write a book, not solely about living with FA, but about my time on this earth thus far, during which FA has played a significant role. The title of my little memoir is *Two in a Million*, which, if I'm correct, is the approximate statistical incidence of Fanconi's in the general population. I'm continuing to work on the book, and my wish is to attempt to get it published some time over the next two years. I hope you will read it when it comes out! And, if anybody knows a good publisher, feel free to e-mail me...!

In a short article that appeared in

The Irish Times a number of years ago, I wrote: "It's funny, but one of the great teachers in life is serious illness.... I can't imagine myself without this illness; I'm sure it has influenced my personality in so many small ways. Without being sarcastic, it does give you an insight into human suffering, and hopefully influences you for the good."

I stand by that sentiment. Fanconi anaemia, like any life-threatening disease, wreaks horrors on individuals who never deserved them. But, it can also be a catalyst toward the profoundly good, as this community of supporting families itself proves. In place of the lacklustre, serious illness can give rise to overwhelming determination and drive; in place of taking life for granted, it can substitute a deep respect for the value of each human being.

I know I shall never be totally liberated from the worries of Fanconi anaemia. Yet, I come back to this newsletter, and I read about others who have struggled through the toughest times of this disease. I can't but be struck by the raw and humble courage of FA patients. I read about how even those who have lost loved ones to FA vow they will be better people because the illness has been part of their lives.

Almost everyone reading my piece will have been affected by Fanconi anaemia in some way. You have suffered and coped, and have been brought to laughter and to tears by this disease. In the hardest periods ahead, you have every reason to stand tall. You are your own inspiration. ♦

FUNDRAISING

The Himmelreich Family Hits the Ground Running

We would like to take this opportunity to introduce ourselves. We are Peter and Tara Himmelreich, and we reside in Pennsylvania with our two daughters, Emily, age 3, and Megan, who just turned one year. Last summer, we became a part of the Fanconi anemia family when our second-born was diagnosed with FA. Despite our overwhelming feelings of sadness, grief, and hopelessness, we quickly became aware of the severity of the disease and the effort we needed to make to help find a cure.

In March, we kicked off our fundraising campaign by hosting our first annual *Beef n' Beer* benefit. We felt confident that our event would be successful, but had no idea that, in one short evening, we would raise almost \$16,000 for the Fund!!! We started by renting a hall, caterer, and a DJ. We sent out invitations to our friends and family and the response was truly overwhelming! Donations were solicited from local businesses and corporations for a raffle and silent auction. We were thrilled with the response we received from the community! "Ask, and you shall receive." (Believe in it!!) The night was truly amazing and lots of fun.

We have already been asked if there will be a *Beef n' Beer* this year. You can count on it! ♦



Tara and Megan Himmelreich

Mont Blanc Again

by Mai Byrne

Living with Fanconi anaemia can be a positive driving force. It has succeeded in driving me, my husband Des, and some friends to the top of Mont Blanc twice in the last ten years!

We have been living with FA for eleven years. Two of our children, Ben (20) and Jess (15), were diagnosed with this insidious life-threatening disorder. Ruth (17) does not have FA. After the initial blinding terror and searing pain of this diagnosis, we determined to be proactive in raising funds to further research toward a cure.

The presence of FA in our lives

generated support among family and friends for a number of fundraising ventures over the past eleven years, from BBQs to bag packs to sponsored cycles and walks. In 1995, we climbed Mont Blanc for the first time and raised around US\$30,000, which was split evenly between the FA Research Fund and Our Lady's Hospital for Sick Children in Crumlin, Dublin, Ireland. When Des suggested in 2004 that we do a second sponsored Mont Blanc climb, but this time undertake a harder route, I protested—too tough, too long, too much organisation! However, Des persisted.

He was right. Calvin Torrans and Ian Rea, qualified mountain guides and friends of ours, agreed to lead us on the "Mont Blanc Traverse" (those of you who know Mont Blanc will understand the difference between Mont Blanc and the Mont Blanc Traverse). Dave Fleming, an employee of Des's, and Ken Byrne, our friend, also agreed to come along. Des was positive, determined, and fit. I was sceptical, wavering, and unfit, but said I'd climb nevertheless. The plan was to invite friends, family and acquaintances to sponsor the "expedition." Des was the driving

continued on page 24

Chris Hull Memorial Sigma Pi Open

by Andy Harner

The early morning of Saturday, June 11, 2005 found 50 fraternity brothers, family members, and friends of the late Christopher Hull lined up in golf carts ready to begin the 6th Annual Chris Hull Memorial Sigma Pi Open. Since losing Chris to his lifelong battle with FA, his fraternity brothers of the Theta Chapter of Sigma Pi Fraternity have annually sponsored a golf tournament in his memory. Funds raised from the tournament benefit the FA Research Fund.

In addition to the tournament registration fees, the fraternity has found unique ways to raise extra money for the FA Research Fund. Over the past few years, week-long stays at golf resorts have been raffled at the event. Another popular fundraiser has been the auctioning of Penn State vs. Ohio State football tickets.

As a freshman at Penn State in the fall of 1984, Chris joined Sigma Pi as a pledge, or associate member. One of the reasons that Chris chose Sigma Pi was that his older brother, Jeff, had also been a Sigma Pi member. Fellow members recall Chris as an active, involved member of the chapter. Favorite memories include Chris as being a “big brother” or formal mentor to a large number of younger men, during their pledge semesters. Chris was a trusted advisor and confidant who encouraged his “little brothers” to work through the long hours and hard work of the pledge period, just as he had done.

During the golf tournament weekend, stories about Chris are always told and re-told, such as the Halloween party where Chris arrived



Chris Hull, far left, with fraternity brothers Andy Harner, Joel Cliff, Scott Bennett, and Curt Spangler and spouses enjoy the day prior to the 1994 Rose Bowl.

as a “Chris”-mas tree, complete with battery-powered twinkle lights, ornaments and pine-green tights. During his years in the fraternity, Chris Hull lived life to the fullest, experiencing all that Penn State and Sigma Pi had to offer. Fraternity members are pleased to have been able to donate over \$25,000 during the past six years to the FA Research Fund, in Chris’ memory. The tournament will return again next year, in early June, to the Penn State Blue golf course in State College, PA. ♦

Use of Logo

A reminder to our FA families: please use our logo or letterhead only after you have consulted the staff of the FA Research Fund and received their approval. This is necessary to be sure our messages are accurate and consistent and helps to avoid legal complications. We are happy to collaborate on fundraisers and mailings.

Charity Navigator Awards the Fund Its Highest Rating

America’s premier independent charity evaluator, Charity Navigator, has awarded the Fanconi Anemia Research Fund its highest rating, *Four Stars*.

Charity Navigator helps charitable givers make intelligent giving decisions by providing in-depth, objective ratings and analysis of the financial health of America’s charities in the areas of fundraising efficiency, fundraising expenses, program expenses, and administrative expenses. According to Charity Navigator, “charities that are efficient spend less money to raise more. Their fundraising efforts stay in line with the scope of the programs and services they provide. They keep administration costs within reasonable limits. They devote the majority of their spending to the programs and services they exist to provide.”

In earning Charity Navigator’s highest *Four Star* rating, the Fund has demonstrated exceptional financial health, outperforming most of its peers in its efforts to manage and increase its financial assets in the most fiscally responsible way possible.

The FA Research Fund is proud that its outstanding record of fiscal responsibility has placed us in this elite *Four Star* group. Administrative costs of the Fund have consistently been far below average for similar organizations. The Fund’s 2004 annual audit documented that its administrative costs were 3.64% and fundraising costs were 2.77%, for a combined total of 6.41%. These results are exemplary by any standard. Donors to the Fund can be confident that their donations will be spent strategically to advance critically-needed FA research. ♦

Coley's Cause Memorial Golf Tournament

by Kristin and Todd Levine

When we lost our precious Nicole ("Coley") in October 2004, we were determined to create an annual Fanconi Anemia fundraising event that could serve two purposes: to bring family and friends together once a year to honor the memory of our beautiful, courageous and spirited daughter, and to raise awareness and much needed research funds for FA. With her birthday set in the heart of golf season, we decided to establish the annual Coley's Cause Memorial Golf Tournament.

Having never organized an event like this, we nevertheless jumped in with both feet and charged forward with the help of many friends and neighbors. Within weeks of announcing the tournament, we had a full field of 144 registered golfers, and the sponsorships and donations were flowing in. Some of the key things we did to make the event a success were:

- Establishing a website (www.coleyscause.com) to inform people about FA and Nicole's journey, as well as to ease registration and sponsorship transactions and to provide a space for sponsor advertisements.
- Enlisting friends to solicit local business through personal contact and/or mass mailings (with information about Nicole, FARE, and sponsorship opportunities).
- Offering multiple levels of sponsorship, from \$75 to \$1,500. We got eight \$1,500 donations!
- Conducting a raffle and silent auction after the tournament.
- Holding on-course "chance" contests such as hit-the-green, long drive, and putting contest.

The end result of this labor of love was a fantastic tournament that raised over \$30,000 for Coley's Cause—of which \$25,000 was donated to FARE. We're hoping for at least a 50% increase in proceeds for next year's tournament. Most impressively, every attendee made a point to tell us to sign him or her up for next year!

We would be happy to talk to anyone who has or is thinking of planning a similar tournament to share more details on our successes and on lessons learned from our inaugural event. ♦



Todd and Kristin Levine with Amy Vangel in the middle.

Fundraising Assistance

Did you know that 85% of the donations to the FA Research Fund are raised by FA families? Obviously, we need the efforts of everyone who reads this newsletter!

The staff of the Fund stands ready to assist you with your fundraising efforts. We'll help you write or edit your fundraising letter; photocopy it; provide the postage; and mail it from our office, using your mailing list. If you're going to hold a fundraising event, we'll provide similar help. Fundraising help is also available through the FA Fundraising Team leader in your area.

The FA Research Fund asks FA parents to make certain that any event they hold is covered by liability insurance. Coverage for a one-time event is often available through a family's homeowners insurance as a relatively inexpensive insurance rider. Please contact the Fund if you need assistance obtaining or paying for this required insurance.

When a donation is received, we'll send a letter of thanks from the Fund with a tax receipt, and we'll notify you that a donation has been made in your name. One request: Please ask your donors to write their donation check to the "Fanconi Anemia Research Fund."

Our sincere thanks go to all of you for your efforts to raise funds to combat this devastating disease.

Family Fundraising Efforts

From January 1 through August 31, 2005, FA families raised \$674,372 for Fanconi anemia research. The Fund also received donations of \$7,681 through the United Way and \$4,993 through the Combined Federal Campaign.

For years, Lynn and Dave Frohnmayer have raised the vast majority of funds to advance FA research, compared to all other FA families (69% vs. 31% in the year 2000, for example). However, by 2004, through the determined efforts of over 100 FA families, that percentage has changed to 45% vs. 55%. **All other FA families now raise slightly more funds than the Frohnmayers!**

We thank all of you who have worked so hard to raise critically needed research dollars while simultaneously spending countless hours dealing with the personal anguish of Fanconi anemia. We are extremely grateful for each effort.

We must continue this momentum to meet our 2005 fundraising goal of \$2.2 million. We're confident that, with your help, we can do so. Members of the staff of the Fund will be happy to help you with your fundraising efforts, as will the leaders of the FA Fundraising Teams.

FA families who have raised funds so far in 2005 are the following:

\$100,000 and up

Dave and Lynn Frohnmayer

Jack and Lisa Nash

Mark and Susan Trager

Mike and Beth Vangel

Rick and Lynn Sablosky

Bob and Andrea Sacks

Bill and Connie Schenone

Irwin and Leona Selden

\$50,000 to \$99,999

John and Audrey Barrow

John and Kim Connelly

Alan and Rachel Grossman

Glen Shearer and Peggy Padden

\$1,000 to \$4,999

Ken and Jeanne Atkinson

Cherie Banks

Darryl Blecher and Diana Fitch

Randy and Nancy Bloxom

Mike and Kerrie Brannock

Donald and Danielle Burkin

David and Kim Chew

Donna DellaRatta

Pat and Mary DiMarino

Antonino and Marie DiMercurio

David and Mary Ann Fiaschetti

Allan Goldberg and Lori Strongin

Andrew and Jennifer Gough

John and Martina Hartmann

Jeff and Beth Janock

Lila Keleher

Erik Kjos-Hanssen and Turid Frislid

Lynette Lowrimore

Fred and Nancy Nunes

Steve Perkins and Karen Magrath

Tyler Morrison and Rachel Altmann

Tony and Lina Nahas

Robert and Mary Nori

Derek and Ginger Persson

John and Dianne Ploetz

Pedro and Marina Ravelo

Marcia Reardon

Up to \$999

Andrew and Vicki Athens

Mark and Linda Baumiller

Roger and Annette Bevelhymer

Paul and Tracy Birchall

Michael and Diane Bradley

Richard Briga

Floyd and Susan Clark

Jeanette Clark

Tyler and Teresa Clifton

Bill and Pat Danks

Dottie Day

Charles and Dahne Deeks

James and Carol Dillon

Nathan and Ann Eckstadt

Gene and Lynn Eddy

Sharon Ellis

Frederic Engel

Wendy Epps

Carol Felmy and Michael Glas

Stephen and Doreen Flynn

Nanette Vannostran Foster

Gary and Melody Ganz

Cindy and Brian Gaudet

\$25,000 to \$49,999

Peter and Tara Himmelreich

Todd and Kristin Levine

\$20,000 to \$24,999

Mark and Diane Pearl

\$15,000 to \$19,999

Jeffrey and Donna Boggs

Tanner and Jessica Lindsay

Des Murnane and Mai Byrne

Family Fundraising Efforts

continued from page 22

Pat and Maria Gleason
Maria Godwin
Michael Greenberg
Sean and Helen Healey
Roger and Eleanor Herman
Jeff and Judy Hoffman
John and Karilyn Kelson
Robert and Jennifer Kiesel
Kristina King
Ayala Laufer
Eugene and Renee Lemmon
Peg LeRoux
Michael and Myra Lewis
Larry and Gayle Kicari
Dan and Nikki McCarthy
Steve and Allison McClay
Kevin and Barbara McKee
Gianna and Lauren Megna
Charles and Cecilia Meloling
Adam and Olivia Mindle
Griff and Cecilia Morgan
John and Betty Mozisek
Sheila Muhlen
Kenny and Lisa Myhan
Louis and Virginia Napoles
Freddy and Kathy Pharris
Tom Plummer
Lynn and Shirley Quilici
Jack and Tannis Redekop
Les and Nancy Ross
Thomas and Brenda Seiford
Bryan and Karen Siebenthal
Jim and Carol Siniawski
Greg and Brandi Stuart
Marc and Sandi Weiner
Marge Zaborney

How You Can Help

Your donations have helped move this fatal disease from an orphaned status in 1989 to a disease with treatments that now buy precious time for FA patients. As the genetic basis of Fanconi anemia continues to be deciphered, your donations are also having an impact on the lives of millions in the general population. We continue to move to the mainstream of scientific interest. To help us in this fight, consider these ways to donate:

Gifts to celebrate an occasion: If you are celebrating a birth, a birthday, an anniversary, a graduation, a marriage or other gift-worthy event, consider asking that donations be made to the Fund in honor of the reason for the event.

Gifts to commemorate a loved one: Families who have lost a loved one may ask that a donation to the FA Research Fund be made in memory of the deceased individual. The Fund has received many thousands of dollars from caring people who have responded to obituary announcements.

Bequests: If you are preparing or reviewing your *Last Will and Testament*, consider making a bequest to the Fund.

Matching Gifts: Many employers will match the charitable gift of an employee. This is an excellent way to double your donation.

Gifts of Appreciated Property: Donors who have property that has gained greatly in value (stock, vacation homes, art items, etc.) can avoid tax liabilities and convey enormous support by gifting this property to the Fund. Please contact us for helpful advice and suggestions.

United Way or Combined Federal Campaign: If you work for an organization covered by either of these organizations, consider making a donation via your workplace and asking your colleagues to do the same.

Donations Online: Look for the PayPal button in the Donations section of our web page (www.fanconi.org)

Donations by Telephone: Call us at (541) 687-4658 or toll free at (800) 828-4891.

Donations by Mail: 1801 Willamette Street, Suite 200, Eugene, OR 97401.

Mont Blanc Again

continued from page 19

force. He organised the invitational letter and sponsorship card, and wrote to all his clients. Dave and Ken were sponsored by their friends and family. I also appealed to companions and work colleagues to provide financial support for my endeavour.

Our motley crew climbed Mont Blanc du Tacul, Mont Maudi, and eventually arrived at the summit of Mont Blanc itself, exhilarated, one morning in August 2004—having walked continuously for 24 hours, leaping over bergshunds, wielding ice axes, and hauling ourselves up steep, relentless snow slopes. You're cold, the altitude makes you sick, and you want to lie in the snow and die. However, the quiet courage of all those who suffer from FA inspired us to continue.

We raised over US\$36,000, and, once again, the funds were evenly divided between the FA Research Fund and Crumlin Hospital, where Drs. Finn Breathnach, Ann O'Meara, and Aengus O'Marcaigh have been our medical saviours. It was in Crumlin, under the direction of Dr. O'Marcaigh, that Ben underwent his matched unrelated bone marrow transplant four years ago.

The novelty of our fundraising extravaganza excited interest and awakened enthusiasm to a degree that might not otherwise have been the case and, thus, generated greater financial support. So, if you have a fundraising idea that seems off the wall, ridiculous, or mad—do it! If those with FA can live their lives in quiet and determined courage, then the rest of us can climb our own Mont Blancs in support. ♦



**Fanconi
Anemia**

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